Heinz Jungbluth

List of Publications by Year
in descending order

154 papers	13,228 citations	34105 52 h-index	23533 111 g-index
166 all docs	166 docs citations	166 times ranked	19760 citing authors

#	Article	IF	Citations
1	Genotype-phenotype correlations in ocular manifestations of Marinescoâ€“SjÃ¶gren syndrome: Case report and literature review. European Journal of Ophthalmology, 2022, 32, NP92-NP97.	1.3	2
2	RYR1-Related Rhabdomyolysis: A Spectrum of Hypermetabolic States Due to Ryanodine Receptor Dysfunction. Current Pharmaceutical Design, 2022, 28, 2-14.	1.9	11
3	The spectrum of neurodevelopmental, neuromuscular and neurodegenerative disorders due to defective autophagy. Autophagy, 2022, 18, 496-517.	9.1	18
4	259th ENMC international workshop: Anaesthesia and neuromuscular disorders 11 December, 2020 and 28â€“29 May, 2021. Neuromuscular Disorders, 2022, 32, 86-97.	0.6	6
5	Referral Indications for Malignant Hyperthermia Susceptibility Diagnostics in Patients without Adverse Anesthetic Events in the Era of Next-generation Sequencing. Anesthesiology, 2022, 136, 940-953.	2.5	12
6	Pre-operative exercise and pyrexia as modifying factors in malignant hyperthermia (MH). Neuromuscular Disorders, 2022, 32, 628-634.	0.6	12
7	Genetic neuropathies presenting with CIDP-like features in childhood. Neuromuscular Disorders, 2021, 31, 113-122.	0.6	6
8	An ancestral 10-bp repeat expansion in VWA1 causes recessive hereditary motor neuropathy. Brain, 2021, 144, 584-600.	7.6	20
9	INPP5K and SIL1 associated pathologies with overlapping clinical phenotypes converge through dysregulation of PHGDH. Brain, 2021, 144, 2427-2442.	7.6	7
10	Spectrum of Clinical Features in X-Linked Myotubular Myopathy Carriers. Neurology, 2021, 97, e501-e512.	1.1	9
11	The neuromuscular and multisystem features of RYR1-related malignant hyperthermia and rhabdomyolysis. Medicine (United States), 2021, 100, e26999.	1.0	8
12	Clinical, genetic, and histological features of centronuclear myopathy in the Netherlands. Clinical Genetics, 2021, 100, 692-702.	2.0	7
13	Making sense of missense variants in TTN-related congenital myopathies. Acta Neuropathologica, 2021, 141, 431-453.	7.7	34
14	The etiology of rhabdomyolysis: an interaction between genetic susceptibility and external triggers. European Journal of Neurology, 2021, 28, 647-659.	3.3	26
15	HyperCKemia and rhabdomyolysis in the neuroleptic malignant and serotonin syndromes: A literature review. Neuromuscular Disorders, 2020, 30, 949-958.	0.6	9
16	rAAV-related therapy fully rescues myonuclear and myofilament function in X-linked myotubular myopathy. Acta Neuropathologica Communications, 2020, 8, 167.	5.2	12
17	<scp>RBCK1</scp>â€related disease: A rare multisystem disorder with polyglucosan storage, autoâ€inflammation, recurrent infections, skeletal, and cardiac myopathyâ€”Four additional patients and a review of the current literature. Journal of Inherited Metabolic Disease, 2020, 43, 1002-1013.	3.6	23
18	The congenital myopathies. , 2020, , 451-461.		0

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19	MYO-MRI diagnostic protocols in genetic myopathies. Neuromuscular Disorders, 2019, 29, 827-841.	0.6	46
20	Therapeutic Aspects in Congenital Myopathies. Seminars in Pediatric Neurology, 2019, 29, 71-82.	2.0	10
21	Impairments in contractility and cytoskeletal organisation cause nuclear defects in nemaline myopathy. Acta Neuropathologica, 2019, 138, 477-495.	7.7	25
22	Transcriptional Regulation of the Glutamate/GABA/Glutamine Cycle in Adult Glia Controls Motor Activity and Seizures in Drosophila. Journal of Neuroscience, 2019, 39, 5269-5283.	3.6	26
23	A novel case of MSTO1 gene related congenital muscular dystrophy with progressive neurological involvement. Neuromuscular Disorders, 2019, 29, 448-455.	0.6	9
24	Recessive MYH7-related myopathy in two families. Neuromuscular Disorders, 2019, 29, 456-467.	0.6	14
25	Aberrant regulation of epigenetic modifiers contributes to the pathogenesis in patients with selenoprotein N â€ related myopathies. Human Mutation, 2019, 40, 962-974.	2.5	13
26	Fatal awake malignant hyperthermia episodes in a family with malignant hyperthermia susceptibility: a case series. Canadian Journal of Anaesthesia, 2019, 66, 540-545.	1.6	16
27	The histopathological spectrum of malignant hyperthermia and rhabdomyolysis due to RYR1 mutations. Journal of Neurology, 2019, 266, 876-887.	3.6	26
28	Driving next-generation autophagy researchers towards translation (DRIVE), an international PhD training program on autophagy. Autophagy, 2019, 15, 347-351.	9.1	4
29	Functional impairments, fatigue and quality of life in RYR1-related myopathies: A questionnaire study. Neuromuscular Disorders, 2019, 29, 30-38.	0.6	20
30	Congenital disorders of autophagy â€“ a novel class of neurological and neuromuscular disorders linking abnormal neurodevelopment and neurodegeneration. Nervenheilkunde, 2019, 38, .	0.0	0
31	Parental mosaicism in RYR1 -related Central Core Disease. Neuromuscular Disorders, 2018, 28, 422-426.	0.6	5
32	Unusual Presentations of Dystrophinopathies in Childhood. Pediatrics, 2018, 141, S510-S514.	2.1	4
33	Neck-Tongue Syndrome: An Underrecognized Childhood Onset Cephalalgia. Journal of Child Neurology, 2018, 33, 347-350.	1.4	6
34	Congenital myopathies: disorders of excitationâ€“contraction coupling and muscle contraction. Nature Reviews Neurology, 2018, 14, 151-167.	10.1	212
35	Atypical periodic paralysis and myalgia. Neurology, 2018, 90, e412-e418.	1.1	39
36	Compound heterozygous RYR1 mutations in a preterm with arthrogryposis multiplex congenita and prenatal CNS bleeding. Neuromuscular Disorders, 2018, 28, 54-58.	0.6	5

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37	The Vici syndrome protein EPG5 regulates intracellular nucleic acid trafficking linking autophagy to innate and adaptive immunity. Autophagy, 2018, 14, 22-37.	9.1	23
38	STAC3variants cause a congenital myopathy with distinctive dysmorphic features and malignant hyperthermia susceptibility. Human Mutation, 2018, 39, 1980-1994.	2.5	42
39	ECEL1 gene related contractural syndrome: Long-term follow-up and update on clinical and pathological aspects. Neuromuscular Disorders, 2018, 28, 741-749.	0.6	15
40	PRUNE1 Deficiency: Expanding the Clinical and Genetic Spectrum. Neuropediatrics, 2018, 49, 330-338.	0.6	11
41	219th ENMC International Workshop Titinopathies International database of titin mutations and phenotypes, Heemskerk, The Netherlands, 29 Aprilâ€“1 May 2016. Neuromuscular Disorders, 2017, 27, 396-407.	0.6	29
42	Myopathology in times of modern imaging. Neuropathology and Applied Neurobiology, 2017, 43, 24-43.	3.2	34
43	Autopsy findings in EPG5â€related Vici syndrome with antenatal onset. American Journal of Medical Genetics, Part A, 2017, 173, 2522-2527.	1.2	6
44	Stall in Canonical Autophagy-Lysosome Pathways Prompts Nucleophagy-Based Nuclear Breakdown in Neurodegeneration. Current Biology, 2017, 27, 3626-3642.e6.	3.9	47
45	Current and future therapeutic approaches to the congenital myopathies. Seminars in Cell and Developmental Biology, 2017, 64, 191-200.	5.0	29
46	Ca2+ handling abnormalities in early-onset muscle diseases: Novel concepts and perspectives. Seminars in Cell and Developmental Biology, 2017, 64, 201-212.	5.0	19
47	Polymyositis without Beneficial Response to Steroid Therapy: Should Miyoshi Myopathy be a Differential Diagnosis?. American Journal of Case Reports, 2017, 18, 17-21.	0.8	9
48	Compound Heterozygous RYR1 Mutation in a Preterm with Arthrogryposis and Core Myopathy with Prenatal CNS Bleedings. Neuropediatrics, 2017, 48, S1-S45.	0.6	0
49	Cellular, biochemical and molecular changes in muscles from patients with X-linked myotubular myopathy due toMTM1mutations. Human Molecular Genetics, 2016, 26, ddw388.	2.9	20
50	Congenital myopathies: not only a paediatric topic. Current Opinion in Neurology, 2016, 29, 642-650.	3.6	37
51	CAV3 mutations causing exercise intolerance, myalgia and rhabdomyolysis: Expanding the phenotypic spectrum of caveolinopathies. Neuromuscular Disorders, 2016, 26, 504-510.	0.6	38
52	Exertional rhabdomyolysis: physiological response or manifestation of an underlying myopathy?. BMJ Open Sport and Exercise Medicine, 2016, 2, e000151.	2.9	73
53	RYR1-related rhabdomyolysis: A common but probably underdiagnosed manifestation of skeletal muscle ryanodine receptor dysfunction. Revue Neurologique, 2016, 172, 546-558.	1.5	28
54	Dantrolene as a possible prophylactic treatment for RYR1â€related rhabdomyolysis. European Journal of Neurology, 2016, 23, e56-7.	3.3	10

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55	An RYR1 mutation associated with malignant hyperthermia is also associated with bleeding abnormalities. Science Signaling, 2016, 9, ra68.	3.6	34
56	Reply: Aberrant splicing induced by the most commonEPG5mutation in an individual with Vici syndrome. Brain, 2016, 139, e53-e53.	7.6	4
57	The Human 343delT HSPB5 Chaperone Associated with Early-onset Skeletal Myopathy Causes Defects in Protein Solubility. Journal of Biological Chemistry, 2016, 291, 14939-14953.	3.4	16
58	Vici syndrome: a review. Orphanet Journal of Rare Diseases, 2016, 11, 21.	2.7	55
59	Downstream effects of plectin mutations in epidermolysis bullosa simplex with muscular dystrophy. Acta Neuropathologica Communications, 2016, 4, 44.	5.2	35
60	Salbutamol-responsive fetal acetylcholine receptor inactivation syndrome. Neurology, 2016, 86, 692-694.	1.1	10
61	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). Autophagy, 2016, 12, 1-222.	9.1	4,701
62	HeterozygousKIDINS220/ARMSnonsense variants cause spastic paraplegia, intellectual disability, nystagmus, and obesity. Human Molecular Genetics, 2016, 25, 2158-2167.	2.9	37
63	Lyme Neuroborreliosis: A Potentially Preventable Cause of Stroke. Journal of Pediatrics, 2016, 170, 334-334.e1.	1.8	8
64	EPG5-related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. Brain, 2016, 139, 765-781.	7.6	99
65	Congenital disorders of autophagy: an emerging novel class of inborn errors of neuro-metabolism. Brain, 2016, 139, 317-337.	7.6	126
66	â€œHuman Stress Syndromeâ€•and the Expanding Spectrum of RYR1-Related Myopathies. Cell Biochemistry and Biophysics, 2016, 74, 85-87.	1.8	14
67	Autophagy â€“ a fundamental cellular mechanism on the verge of clinical translation. Neuropathology and Applied Neurobiology, 2015, 41, 598-600.	3.2	1
68	Goldbergâ€“Shprintzen megacolon syndrome with associated sensory motor axonal neuropathy. American Journal of Medical Genetics, Part A, 2015, 167, 1300-1304.	1.2	23
69	Rhabdomyolysis: a genetic perspective. Orphanet Journal of Rare Diseases, 2015, 10, 51.	2.7	101
70	<scp>RYR</scp>1â€related myopathies: a wide spectrum of phenotypes throughout life. European Journal of Neurology, 2015, 22, 1094-1112.	3.3	111
71	Recessive truncating IGHMBP2 mutations presenting as axonal sensorimotor neuropathy. Neurology, 2015, 84, 523-531.	1.1	22
72	Fetal acetylcholine receptor inactivation syndrome. Neurology: Neuroimmunology and NeuroInflammation, 2015, 2, e57.	6.0	50

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73	Early-onset movement disorder and epileptic encephalopathy due to de novo dominant SCN8A mutation. Seizure: the Journal of the British Epilepsy Association, 2015, 26, 69-71.	2.0	26
74	Epigenetic changes as a common trigger of muscle weakness in congenital myopathies. Human Molecular Genetics, 2015, 24, 4636-4647.	2.9	44
75	RYR1-related malignant hyperthermia with marked cerebellar involvement â€“ A paradigm of heat-induced CNS injury?. Neuromuscular Disorders, 2015, 25, 138-140.	0.6	21
76	Congenital myopathies. Neurology, 2015, 84, 28-35.	1.1	106
77	SIL1-related Marinescoâ€“Sjoegren syndrome (MSS) with associated motor neuronopathy and bradykinetic movement disorder. Neuromuscular Disorders, 2015, 25, 585-588.	0.6	14
78	The Congenital Myopathies. , 2015, , 1121-1129.		2
79	Compound RYR1 heterozygosity resulting in a complex phenotype of malignant hyperthermia susceptibility and a core myopathy. Neuromuscular Disorders, 2015, 25, 567-576.	0.6	28
80	The neuromuscular differential diagnosis of joint hypermobility. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2015, 169, 23-42.	1.6	22
81	Severe Central Sleep Apnea in Vici Syndrome. Pediatrics, 2015, 136, e1390-e1394.	2.1	9
82	Pathogenic Mechanisms in Centronuclear Myopathies. Frontiers in Aging Neuroscience, 2014, 6, 339.	3.4	89
83	Clinical utility gene card for: Vici Syndrome. European Journal of Human Genetics, 2014, 22, 435-435.	2.8	13
84	Mutations in SCN4A: A Rare but Treatable Cause of Recurrent Life-Threatening Laryngospasm. Pediatrics, 2014, 134, e1447-e1450.	2.1	34
85	Novel Mutations Widen the Phenotypic Spectrum of Slow Skeletal/Î²-Cardiac Myosin (MYH7) Distal Myopathy. Human Mutation, 2014, 35, 868-879.	2.5	79
86	G.P.22. Neuromuscular Disorders, 2014, 24, 801.	0.6	3
87	Outcome of children with acetylcholine receptor (AChR) antibody positive juvenile myasthenia gravis following thymectomy. Neuromuscular Disorders, 2014, 24, 25-30.	0.6	24
88	Approach to the diagnosis of congenital myopathies. Neuromuscular Disorders, 2014, 24, 97-116.	0.6	239
89	Ophthalmologic Features of Vici Syndrome. Journal of Pediatric Ophthalmology and Strabismus, 2014, 51, 214-220.	0.7	15
90	Congenital (Structural) Myopathies. , 2013, , 1-51.		0

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91	A novel late-onset axial myopathy associated with mutations in the skeletal muscle ryanodine receptor (RYR1) gene. Journal of Neurology, 2013, 260, 1504-1510.	3.6	71
92	RyR1 Deficiency in Congenital Myopathies Disrupts Excitation-Contraction Coupling. Human Mutation, 2013, 34, 986-996.	2.5	40
93	Congenital myopathy with focal loss of cross-striations revisited. Neuromuscular Disorders, 2013, 23, 160-164.	0.6	1
94	198th ENMC International Workshop: 7th Workshop on Centronuclear (Myotubular) myopathies, 31st May â€“ 2nd June 2013, Naarden, The Netherlands. Neuromuscular Disorders, 2013, 23, 1033-1043.	0.6	14
95	Mutations in RYR1 are a common cause of exertional myalgia and rhabdomyolysis. Neuromuscular Disorders, 2013, 23, 540-548.	0.6	169
96	Congenital myopathies â€“ Clinical features and frequency of individual subtypes diagnosed over a 5-year period in the United Kingdom. Neuromuscular Disorders, 2013, 23, 195-205.	0.6	113
97	Novel deletion of lysine 7 expands the clinical, histopathological and genetic spectrum of TPM2-related myopathies. Brain, 2013, 136, 508-521.	7.6	53
98	Recessive mutations in EPG5 cause Vici syndrome, a multisystem disorder with defective autophagy. Nature Genetics, 2013, 45, 83-87.	21.4	231
99	Clinical utility gene card for: Central core disease. European Journal of Human Genetics, 2012, 20, 5-5.	2.8	4
100	Clinical utility gene card for: Multi-minicore disease. European Journal of Human Genetics, 2012, 20, 5-5.	2.8	2
101	Consensus Statement on Standard of Care for Congenital Myopathies. Journal of Child Neurology, 2012, 27, 363-382.	1.4	147
102	X-linked myotubular myopathy due to a complex rearrangement involving a duplication of MTM1 exon 10. Neuromuscular Disorders, 2012, 22, 384-388.	0.6	11
103	Myopathic causes of exercise intolerance with rhabdomyolysis. Developmental Medicine and Child Neurology, 2012, 54, 886-891.	2.1	54
104	Mutations in MYH7 cause Multi-minicore Disease (MmD) with variable cardiac involvement. Neuromuscular Disorders, 2012, 22, 1096-1104.	0.6	73
105	Clinical utility gene card for: Centronuclear and myotubular myopathies. European Journal of Human Genetics, 2012, 20, 1101-1101.	2.8	28
106	Clinical and genetic findings in a large cohort of patients with ryanodine receptor 1 gene-associated myopathies. Human Mutation, 2012, 33, 981-988.	2.5	145
107	Impaired neuromuscular transmission and response to acetylcholinesterase inhibitors in centronuclear myopathies. Neuromuscular Disorders, 2011, 21, 379-386.	0.6	96
108	Kingâ€“Denborough syndrome with and without mutations in the skeletal muscle ryanodine receptor (RYR1) gene. Neuromuscular Disorders, 2011, 21, 420-427.	0.6	97

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109	Core Myopathies. Seminars in Pediatric Neurology, 2011, 18, 239-249.	2.0	120
110	Enhanced excitation-coupled Ca2+ entry induces nuclear translocation of NFAT and contributes to IL-6 release from myotubes from patients with central core disease. Human Molecular Genetics, 2011, 20, 589-600.	2.9	22
111	The use of rituximab in myasthenia gravis and Lambert-Eaton myasthenic syndrome. Journal of Neurology, Neurosurgery and Psychiatry, 2011, 82, 671-673.	1.9	107
112	Muscle Magnetic Resonance Imaging in Congenital Myopathies Due to Ryanodine Receptor Type 1 Gene Mutations. Archives of Neurology, 2011, 68, 1171.	4.5	89
113	Muscle histology vs MRI in Duchenne muscular dystrophy. Neurology, 2011, 76, 346-353.	1.1	134
114	Vici syndrome associated with sensorineural hearing loss and evidence of neuromuscular involvement on muscle biopsy. American Journal of Medical Genetics, Part A, 2010, 152A, 741-747.	1.2	40
115	RYR1 mutations are a common cause of congenital myopathies with central nuclei. Annals of Neurology, 2010, 68, 717-726.	5.3	230
116	Centronuclear myopathy with cataracts due to a novel dynamin 2 (DNM2) mutation. Neuromuscular Disorders, 2010, 20, 49-52.	0.6	37
117	Multi-minicore disease and atypical periodic paralysis associated with novel mutations in the skeletal muscle ryanodine receptor (RYR1) gene. Neuromuscular Disorders, 2010, 20, 166-173.	0.6	78
118	A recessive ryanodine receptor 1 mutation in a CCD patient increases channel activity. Cell Calcium, 2009, 45, 192-197.	2.4	16
119	Joint hypermobility as a distinctive feature in the differential diagnosis of myopathies. Journal of Neurology, 2009, 256, 13-27.	3.6	47
120	The phenotype of Charcotâ€“Marieâ€“Tooth disease type 4C due to SH3TC2 mutations and possible predisposition to an inflammatory neuropathy. Neuromuscular Disorders, 2009, 19, 264-269.	0.6	78
121	Late-onset axial myopathy with cores due to a novel heterozygous dominant mutation in the skeletal muscle ryanodine receptor (RYR1) gene. Neuromuscular Disorders, 2009, 19, 344-347.	0.6	103
122	Congenital Myasthenic Syndromes in childhood: Diagnostic and management challenges. Journal of Neuroimmunology, 2008, 201-202, 6-12.	2.3	114
123	Centronuclear (myotubular) myopathy. Orphanet Journal of Rare Diseases, 2008, 3, 26.	2.7	267
124	Muscle MRI findings in siblings with juvenile-onset acid maltase deficiency (Pompe disease). Neuromuscular Disorders, 2008, 18, 408-409.	0.6	24
125	Congenital muscle disorders with cores: the ryanodine receptor calcium channel paradigm. Current Opinion in Pharmacology, 2008, 8, 319-326.	3.5	149
126	Molecular mechanisms and phenotypic variation in RYR1-related congenital myopathies. Brain, 2007, 130, 2024-2036.	7.6	161

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127	MRI in DNM2-related centronuclear myopathy: Evidence for highly selective muscle involvement. Neuromuscular Disorders, 2007, 17, 28-32.	0.6	64
128	Centronuclear myopathy due to a de novo dominant mutation in the skeletal muscle ryanodine receptor (RYR1) gene. Neuromuscular Disorders, 2007, 17, 338-345.	0.6	105
129	Central core disease. Orphanet Journal of Rare Diseases, 2007, 2, 25.	2.7	140
130	Multi-minicore Disease. Orphanet Journal of Rare Diseases, 2007, 2, 31.	2.7	114
131	Functional effects of mutations identified in patients with Multiminicore disease. IUBMB Life, 2007, 59, 14-20.	3.4	30
132	Epigenetic Allele Silencing Unveils Recessive RYR1 Mutations in Core Myopathies. American Journal of Human Genetics, 2006, 79, 859-868.	6.2	111
133	Functional properties of ryanodine receptors carrying three amino acid substitutions identified in patients affected by multi-minicore disease and central core disease, expressed in immortalized lymphocytes. Biochemical Journal, 2006, 395, 259-266.	3.7	59
134	Characterization of recessive RYR1 mutations in core myopathies. Human Molecular Genetics, 2006, 15, 2791-2803.	2.9	103
135	Minicore myopathy with ophthalmoplegia caused by mutations in the ryanodine receptor type 1 gene. Neurology, 2005, 65, 1930-1935.	1.1	131
136	Pilot Trial of Salbutamol in Central Core and Multi-Minicore Diseases. Neuropediatrics, 2004, 35, 262-266.	0.6	55
137	Magnetic resonance imaging of muscle in nemaline myopathy. Neuromuscular Disorders, 2004, 14, 779-784.	0.6	98
138	Magnetic resonance imaging of muscle in congenital myopathies associated with RYR1 mutations. Neuromuscular Disorders, 2004, 14, 785-790.	0.6	135
139	What's new in neuromuscular disorders? The congenital myopathies. European Journal of Paediatric Neurology, 2003, 7, 23-30.	1.6	47
140	Early and severe presentation of X-linked myotubular myopathy in a girl with skewed X-inactivation. Neuromuscular Disorders, 2003, 13, 55-59.	0.6	48
141	Principal mutation hotspot for central core disease and related myopathies in the C-terminal transmembrane region of the RYR1 gene. Neuromuscular Disorders, 2003, 13, 151-157.	0.6	118
142	X-inactivation patterns in carriers of X-linked myotubular myopathy. Neuromuscular Disorders, 2003, 13, 468-471.	0.6	40
143	Autosomal recessive inheritance of RYR1 mutations in a congenital myopathy with cores. Neurology, 2002, 59, 284-287.	1.1	157
144	Selective Muscle Involvement on Magnetic Resonance Imaging in Autosomal Dominant Emery-Dreifuss Muscular Dystrophy. Neuropediatrics, 2002, 33, 10-14.	0.6	101

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145	Mutations of the Selenoprotein N Gene, Which Is Implicated in Rigid Spine Muscular Dystrophy, Cause the Classical Phenotype of Multiminicore Disease: Reassessing the Nosology of Early-Onset Myopathies. American Journal of Human Genetics, 2002, 71, 739-749.	6.2	326
146	A recessive form of central core disease, transiently presenting as multi-minicore disease, is associated with a homozygous mutation in the ryanodine receptor type 1 gene. Annals of Neurology, 2002, 51, 750-759.	5.3	181
147	A short protocol for muscle MRI in children with muscular dystrophies. European Journal of Paediatric Neurology, 2002, 6, 305-307.	1.6	105
148	Muscle MRI findings in a three-generation family affected by Bethlem myopathy. European Journal of Paediatric Neurology, 2002, 6, 309-314.	1.6	34
149	Mild phenotype of nemaline myopathy with sleep hypoventilation due to a mutation in the skeletal muscle Î±-actin (ACTA1) gene. Neuromuscular Disorders, 2001, 11, 35-40.	0.6	92
150	Compound heterozygosity and nonsense mutations in the Î±1-subunit of the inhibitory glycine receptor in hyperekplexia. Human Genetics, 2001, 109, 267-270.	3.8	72
151	An unusual case of hyperekplexia. European Journal of Paediatric Neurology, 2000, 4, 77-80.	1.6	7
152	Generalized calcification in a case of dermatomyositis. Neuromuscular Disorders, 2000, 10, 150.	0.6	1
153	Minicore myopathy in children: a clinical and histopathological study of 19 cases. Neuromuscular Disorders, 2000, 10, 264-273.	0.6	60
154	Novel phosphopantothenoylcysteine synthetase (<scp> PPCS </scp>) mutations with prominent neuromuscular features: Expanding the phenotypical spectrum of <scp> PPCS </scp> â€related disorders. American Journal of Medical Genetics, Part A, 0, , .	1.2	1